

Parents' Guide to Hemoglobin E Disease

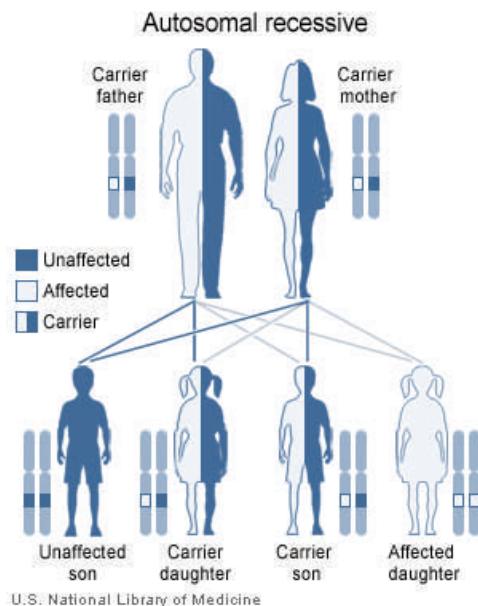
You have just learned that your infant has hemoglobin E disease. Naturally you are concerned and have many questions. This information sheet will help answer some of your questions. However, it should not take the place of an informed discussion with your baby's doctor (primary care provider).

What is Hemoglobin?

Hemoglobin is a protein in the red blood cells. It carries oxygen to all parts of the body and gives blood its red color. There are many hemoglobin types (this is not the same as a blood type). Hemoglobin is inherited through genes, one from each parent. Most people have hemoglobin A, also called adult or normal hemoglobin. The presence of hemoglobin A makes the red blood cells smooth and round. These cells move easily through the blood vessels and deliver oxygen normally to the body.

What is Hemoglobin E Disease?

Hemoglobin E in the red blood cells is responsible for causing hemoglobin E disease. Children *inherit* this disease from their parents as an recessive genetic disorder. This means a hemoglobin E gene is passed from *both* mom and dad to the baby causing hemoglobin E disease (hemoglobin EE). When both parents have one hemoglobin E gene, there is a 1 in 4 or 25% chance with each pregnancy that an infant will inherit two hemoglobin E genes. Persons with hemoglobin EE have red blood cells that are smaller than usual. There are no associated health problems with hemoglobin EE, but the gene for hemoglobin E is passed on to each of one of your future grandchildren. Hemoglobin E disease is not contagious.



What is Hemoglobin E/Beta-Thalassemia Disease?

Children with hemoglobin E/beta-thalassemia *inherit* one gene for hemoglobin E from one parent and one beta-thalassemia gene from the other parent. The beta-thalassemia gene causes the body to make less than the usual amount of hemoglobin. The combination of hemoglobin E/beta-thalassemia is a disease that can be life threatening. Persons with this disease will need special medical care throughout their life.

How Common are Hemoglobin E and Beta-Thalassemia?

The highest frequency of hemoglobin E is among people of Southeast Asian descent, especially those living in or have ancestors from Cambodia, Laos and Thailand. Hemoglobin E is also found in people who live in Vietnam, Malaysia, northeastern India, Bangladesh, Pakistan, Nepal and Sri Lanka and their descendants. Beta-thalassemia is highest among people living in Mediterranean countries and their descendants, such as Greece and Italy. Other areas for beta-thalassemia include the Arabian Peninsula, Turkey, Iran, Africa , India, Southeast Asia and southern China.

What are the Signs and Symptoms of Hemoglobin E/Beta-Thalassemia?

Hemoglobin E/beta-thalassemia disease causes severe destruction of red blood cells. If this condition is left untreated, severe anemia occurs. Other problems are heart failure, an enlarged liver and spleen, poor growth and changes in the bones.

What can be Done to Treat Hemoglobin E/Beta-Thalassemia Disease?

There is no known cure for hemoglobin E/beta-thalassemia. Treatment for this disease includes repeated blood transfusions.

What are the Chances of having a Child with Hemoglobin E/Beta-Thalassemia?

When one parent has hemoglobin E trait (one hemoglobin E gene and one hemoglobin A gene) and the other parent has beta-thalassemia trait (one hemoglobin beta-thalassemia gene and one hemoglobin A gene), there is 1 in 4 or 25% chance with each pregnancy that your child will inherit:

- two hemoglobin A (normal) genes
 - or
- one hemoglobin A gene and one hemoglobin E gene
 - or
- one hemoglobin A gene and one beta-thalassemia gene
 - or
- one hemoglobin E gene and one beta-thalassemia gene

What are the Most Important Things to Remember with Hemoglobin E/Beta-Thalassemia ?

- Work closely with your child's doctor and hematologist (a doctor who is a blood specialist). Make sure your child has regular checkups with them.
- Call your child's doctor when you have questions and have your child seen if you have any medical concerns.

How Do I Get More Information?

Talk with your baby's doctor. You may also want to have a genetic consultation for you and your family to see how these diseases might affect future children or grandchildren.

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